



## NEU1 gene

neuraminidase 1

### Normal Function

The *NEU1* gene provides instructions for making an enzyme called neuraminidase 1 (*NEU1*), which is found in lysosomes. Lysosomes are compartments within cells that use enzymes to digest and recycle materials. The *NEU1* enzyme helps break down large sugar molecules (oligosaccharides) attached to certain proteins (glycoproteins) by removing a substance known as sialic acid.

### Health Conditions Related to Genetic Changes

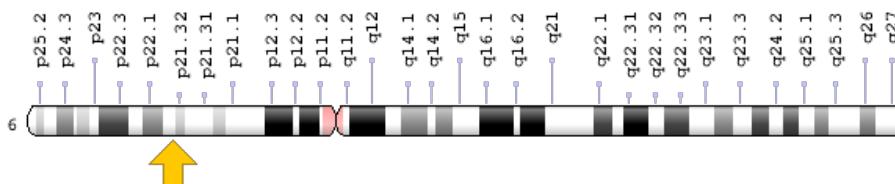
#### sialidosis

At least 42 mutations in the *NEU1* gene have been found to cause sialidosis. Most of these mutations change single protein building blocks (amino acids) used to make the *NEU1* enzyme. Mutations in the *NEU1* gene lead to a shortage (deficiency) of the *NEU1* enzyme. When this enzyme is lacking, large molecules that are usually broken down by the *NEU1* enzyme accumulate inside lysosomes. Conditions such as sialidosis that cause large molecules to build up inside lysosomes are called lysosomal storage disorders. Mutations that eliminate *NEU1* enzyme activity cause more severe signs and symptoms than those that result in some functional enzyme. It is unclear exactly how the accumulation of large molecules within lysosomes leads to the signs and symptoms of sialidosis.

## Chromosomal Location

Cytogenetic Location: 6p21.33, which is the short (p) arm of chromosome 6 at position 21.33

Molecular Location: base pairs 31,859,052 to 31,862,932 on chromosome 6 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

## Other Names for This Gene

- acetylneuraminyl hydrolase
- exo-alpha-sialidase
- FLJ93471
- G9 sialidase
- lysosomal sialidase
- N-acetyl-alpha-neuraminidase 1
- NANH
- NEU
- NEUR1\_HUMAN
- neuraminidase 1 (lysosomal sialidase)
- neuraminidase 1 precursor
- SIAL1
- sialidase 1
- sialidase 1 (lysosomal sialidase)

## Additional Information & Resources

### Educational Resources

- Essentials of Glycobiology (second edition, 2008): Sialic Acids  
<https://www.ncbi.nlm.nih.gov/books/NBK1920/>

## Scientific Articles on PubMed

- PubMed  
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28NEU1%5BTIAB%5D%29+OR+%28neuraminidase+1%5BTIAB%5D%29+OR+%28lysosomal+sialidase%5BTIAB%5D%29%29+AND+%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+720+days%22%5Bdp%5D>

## OMIM

- NEURAMINIDASE 1  
<http://omim.org/entry/608272>

## Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology  
[http://atlasgeneticsoncology.org/Genes/GC\\_NEU1.html](http://atlasgeneticsoncology.org/Genes/GC_NEU1.html)
- ClinVar  
<https://www.ncbi.nlm.nih.gov/clinvar?term=NEU1%5Bgene%5D>
- HGNC Gene Symbol Report  
[http://www.genenames.org/cgi-bin/gene\\_symbol\\_report?q=data/hgnc\\_data.php&hgnc\\_id=7758](http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=7758)
- NCBI Gene  
<https://www.ncbi.nlm.nih.gov/gene/4758>
- UniProt  
<http://www.uniprot.org/uniprot/Q99519>

## **Sources for This Summary**

- Caciotti A, Di Rocco M, Filocamo M, Grossi S, Traverso F, d'Azzo A, Cavicchi C, Messeri A, Guerrini R, Zammarchi E, Donati MA, Morrone A. Type II sialidosis: review of the clinical spectrum and identification of a new splicing defect with chitotriosidase assessment in two patients. *J Neurol.* 2009 Nov;256(11):1911-5. doi: 10.1007/s00415-009-5213-4. Epub 2009 Jul 1. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19568825>
- OMIM: NEURAMINIDASE 1  
<http://omim.org/entry/608272>
- Pattison S, Pankarican M, Rupar CA, Graham FL, Igدورا SA. Five novel mutations in the lysosomal sialidase gene (NEU1) in type II sialidosis patients and assessment of their impact on enzyme activity and intracellular targeting using adenovirus-mediated expression. *Hum Mutat.* 2004 Jan;23(1):32-9.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/14695530>
- Seyrantepe V, Poupetova H, Froissart R, Zabot MT, Maire I, Pshezhetsky AV. Molecular pathology of NEU1 gene in sialidosis. *Hum Mutat.* 2003 Nov;22(5):343-52. Review.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/14517945>

- Wu X, Steigelman KA, Bonten E, Hu H, He W, Ren T, Zuo J, d'Azzo A. Vacuolization and alterations of lysosomal membrane proteins in cochlear marginal cells contribute to hearing loss in neuraminidase 1-deficient mice. *Biochim Biophys Acta*. 2010 Feb;1802(2):259-68. doi: 10.1016/j.bbadi.2009.10.008. Epub 2009 Oct 24.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/19857571>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2818351/>
- Yogalingam G, Bonten EJ, van de Vlekkert D, Hu H, Moshiach S, Connell SA, d'Azzo A. Neuraminidase 1 is a negative regulator of lysosomal exocytosis. *Dev Cell*. 2008 Jul;15(1):74-86. doi: 10.1016/j.devcel.2008.05.005.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/18606142>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2664108/>
- Zanoteli E, van de Vlekkert D, Bonten EJ, Hu H, Mann L, Gomero EM, Harris AJ, Ghersi G, d'Azzo A. Muscle degeneration in neuraminidase 1-deficient mice results from infiltration of the muscle fibers by expanded connective tissue. *Biochim Biophys Acta*. 2010 Jul-Aug;1802(7-8):659-72. doi: 10.1016/j.bbadi.2010.04.002. Epub 2010 Apr 11.  
*Citation on PubMed:* <https://www.ncbi.nlm.nih.gov/pubmed/20388541>  
*Free article on PubMed Central:* <https://www.ncbi.nlm.nih.gov/pmc/articles/PMC2906380/>

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